

UTHOU-16UTL final.ST25 SEQUENCE LISTING

<110> Board of Regents of the University of Texas System MUTATIONS IN A NOVEL PHOTORECEPTOR-PINEAL GENE ON 17P CAUSE LEBER <120> CONGENITAL AMAUROSIS (LCA4) <130> 96606/16UTL <140> 09/765.061 <141> 2001-01-17 <150> 60/331362 <151> 2001-01-14 <160> 78 <170> PatentIn version 3.2 <210> 6689 <211> <212> DNA <213> Homo sapiens <220> <221> gene <222> (1)...(6689)The AIPL1 gene produces the aryl-hydrocarbon receptor interacting <223> protein-like 1 <220> <221> misc_feature (1897)..(1906)<222> <223> n is a, c, g, or t <220> <221> misc_feature (3946)..(3946)<222> <223> n is a, c, g, or t <400> ggcctcccaa agtgctggat tacaggcgtg agtcaccgcg cctggtcccc tgtcttcttt 60 aagaaagctc agcggacctt tttccttctt ggggtggaac aaaaagccaa atctagcaca 120 accctgggca ggggcccaga atcactggaa gcaaaggtgg atgggatagg aggcgaggct 180 gcctgtggac cacaggcccg gcccgagtgg ctctgatgag aagccggggc gcctaggtca 240 ccgccccac cgtctgccct tcccccact cctcctggct gggtaaatcc cagagtctca 300 360 gccgcctaag tgtcttcccc ggaggtgaga ttatctccgc ctgtgctgga cacctccctt tctcctgcag ccatggatgc cgctctgctc ctgaacgtgg aaggggtcaa gaaaaccatt 420 ctgcacgggg gcacgggcga gctcccaaac ttcatcaccg gatcccqagt qaqtqqqqcc 480 cctccggagc agacagggtc ccccacagca gctttcaaca ttccaggtgt gccccaaggc 540 actgtaaaca gctttcagct gtgccaaaaa aacagccagg cagccccagc gctgggcctc 600 cggggagctc ccagcgttta cccattcagg gggcattttt ggtactttgc agattcaact 660 Page 1

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The AIPL1 gene produces the aryl-hydrocarbon receptor interacting protein-like 1

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<223> The AIPL1 gene produces the aryl-hydrocarbon receptor interacting protein-like 1

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The AIPL1 gene produces the aryl-hydrocarbon receptor interacting protein-like 1

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Asn Met Phe Lys Leu Glu Val Trp Glu Ile Leu Leu Thr Ser Met Arg
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                                          Page 26
```

Val His Glu Val Ala Glu Phe Trp Cys Asp Thr Ile His Thr Gly Val 85 90 95 Tyr Pro Ile Leu Ser Arg Ser Leu Arg Gln Met Ala Gln Gly Lys Asp 100 105 110 Pro Thr Glu Trp His Val His Thr Cys Gly Leu Ala Asn Met Phe Ala 115 120 125 His Thr Leu Gly Tyr Glu Asp Leu Asp Glu Leu Gln Lys Glu Pro 130 135 140 Gln Pro Leu Val Phe Val Ile Glu Leu Leu Gln Val Asp Ala Pro Ser Asp Tyr Gln Arg Glu Thr Trp Asn Leu Ser Asn His Glu Lys Met Lys 165 170 175 Ala Val Pro Val Leu His Gly Glu Gly Asn Arg Leu Phe Lys Leu Gly 180 185 190 Arg Tyr Glu Glu Ala Ser Ser Lys Tyr Gln Glu Ala Ile Ile Cys Leu 195 200 205 Arg Asn Leu Gln Thr Lys Glu Lys Pro Trp Glu Val Gln Trp Leu Lys 210 220 Leu Glu Lys Met Ile Asn Thr Leu Ile Leu Asn Tyr Cys Gln Cys Leu 225 230 235 240 Leu Lys Lys Glu Glu Tyr Tyr Glu Val Leu Glu His Thr Ser Asp Ile 245 250 255 Leu Arg His His Pro Gly Ile Val Lys Ala Tyr Tyr Val Arg Ala Arg 260 265 270 Ala His Ala Glu Val Trp Asn Glu Ala Glu Ala Lys Ala Asp Leu Gln 275 280 285 Lys Val Leu Glu Leu Glu Pro Ser Met Gln Lys Ala Val Arg Arg Glu 290 295 300 Leu Arg Leu Leu Glu Asn Arg Met Ala Glu Lys Gln Glu Glu Arg 305 310 315 320 Leu Xaa Cys Arg Asn Met Leu Ser Gln Gly Ala Thr Gln Pro Pro Ala

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Glu Pro Pro Thr Glu Pro Pro Ala Gln Ser Ser Thr Glu Pro Pro Ala 345 350

Glu Pro Pro Thr Ala Pro Ser Ala Glu Leu Ser Ala Gly Pro Pro Ala 355 360 365

Glu Pro Ala Thr Glu Pro Pro Pro Ser Pro Gly His Ser Leu Gln His 370 375

<210> 73

<211> 384

<212> PRT <213> Pan troglodytes

<220>

<221> Peptide

<222> (1)..(384)

<223> Chimpansee AIPL1 Protein

<400> 73

Met Asp Ala Ala Leu Leu Leu Asn Val Glu Gly Val Lys Lys Thr Ile 1 5 10

Leu His Gly Gly Thr Gly Glu Leu Pro Asn Phe Ile Thr Gly Ser Arg 20 25 30

Val Ile Phe His Phe Arg Thr Met Lys Cys Asp Glu Glu Arg Thr Val 35 40 45

Asn Met Phe Lys Leu Glu Val Trp Glu Ile Leu Leu Thr Ser Met Arg 65 70 75

Val His Glu Val Ala Glu Phe Trp Cys Asp Thr Ile His Thr Gly Val 85 90 95

Tyr Pro Ile Leu Ser Arg Ser Leu Arg Gln Met Ala Gln Gly Lys Asp 100 105 110

Pro Thr Glu Trp His Val His Thr Cys Gly Leu Ala Asn Met Phe Ala 115 120 125

Tyr His Thr Leu Gly Tyr Glu Asp Leu Asp Glu Leu Gln Lys Glu Pro 130 135 140

Gln Pro Leu Val Phe Val Ile Glu Leu Leu Gln Val Asp Ala Pro Ser 145 150 155 160

Asp Tyr Gln Arg Glu Thr Trp Asn Leu Ser Asn His Glu Lys Met Lys 165 170 175

Ala Val Pro Val Leu His Gly Glu Gly Asn Arg Leu Phe Lys Leu Gly 180 185 190

Arg Tyr Glu Glu Ala Ser Ser Lys Tyr Gln Glu Ala Ile Ile Cys Leu 195 200 205

Arg Asn Leu Gln Thr Lys Glu Lys Pro Trp Glu Val Gln Trp Leu Lys 210 215 220

Leu Glu Lys Met Ile Asn Thr Leu Ile Leu Asn Tyr Cys Gln Cys Leu 235 240

Leu Lys Lys Glu Glu Tyr Tyr Glu Val Leu Glu His Thr Ser Asp Ile 245 250 255

Leu Arg His His Pro Gly Ile Val Lys Ala Tyr Tyr Val Arg Ala Arg 260 265 270

Ala His Ala Glu Val Trp Asn Glu Ala Glu Ala Lys Ala Asp Leu Arg 275 280 285

Lys Val Leu Glu Leu Glu Pro Ser Met Gln Lys Ala Val Arg Arg Glu 290 295 300

Leu Arg Leu Leu Glu Asn Arg Met Ala Glu Lys Gln Glu Glu Arg 305 310 315 320

Leu Arg Cys Arg Asn Met Leu Ser Gln Gly Ala Thr Gln Pro Pro Ala 325 330 335

Glu Pro Pro Thr Glu Pro Pro Ala Gln Ser Ser Thr Glu Pro Pro Ala 340 345 350

Glu Pro Pro Pro Ala Pro Ser Ala Glu Leu Ser Ala Gly Pro Pro Ala 355 360 365

Glu Thr Ala Thr Glu Pro Pro Pro Ser Pro Gly His Ser Leu Gln His 370 375 380

<210> 74 <211> 372

<212> PRT

<213> Papio anubis

<220>

<221> peptide

<222> (1) .. (372)

<223> Baboon AIPL1 Protein

<400> 74

Met Asp Ala Ala Leu Leu Asn Val Glu Gly Val Lys Lys Thr Ile

5 10 15

Leu His Gly Gly Thr Gly Glu Leu Pro Asn Phe Ile Thr Gly Ser Arg 20 25 30

Val Ile Phe His Phe Arg Thr Met Lys Cys Asp Glu Glu Arg Thr Val 35 40 45

Ile Asp Asp Ser Arg Gln Val Asp Gln Pro Met His Ile Ile Gly
50 55 60

Asn Met Phe Lys Leu Glu Val Trp Glu Ile Leu Leu Thr Ser Met Arg 70 75 80

Val His Glu Val Ala Glu Phe Trp Cys Asp Thr Ile His Thr Gly Val 85 90 95

Tyr Pro Ile Leu Ser Arg Ser Leu Arg Gln Met Ala Gln Gly Lys Asp 100 105 110

Pro Thr Glu Trp His Val His Thr Cys Gly Leu Ala Asn Met Phe Ala 115 120 125

Tyr His Thr Leu Gly Tyr Glu Asp Leu Asp Glu Leu Gln Lys Glu Pro 130 135 140

Gln Pro Leu Ile Phe Val Ile Glu Leu Leu Gln Val Asp Ala Pro Ser 145 150 155 160

Asp Tyr Gln Arg Glu Thr Trp Asn Leu Ser Asn His Glu Lys Met Lys 165 170 175

Val Val Pro Val Leu His Gly Glu Gly Asn Arg Leu Phe Lys Leu Gly 180 185 190

Arg Tyr Glu Glu Ala Ser Ser Lys Tyr Gln Glu Ala Ile Ile Cys Leu 195 200 205 UTHou-16UTL final.ST25
Arg Asn Leu Gln Thr Lys Glu Lys Pro Trp Glu Val Gln Trp Leu Lys
210 215 220 Leu Glu Lys Met Ile Asn Thr Leu Thr Leu Asn Tyr Cys Gln Cys Leu 225 230 235 240 Leu Lys Lys Glu Glu Tyr Tyr Glu Val Leu Glu His Thr Ser Asp Ile 245 250 255 Leu Arg His His Pro Gly Ile Val Lys Ala Tyr Tyr Val Arg Ala Arg 260 265 270 Ala His Ala Glu Val Trp Asn Glu Ala Glu Ala Lys Ala Asp Leu Gln 275 280 285 Lys Val Leu Glu Leu Glu Pro Ser Met Gln Lys Ala Val Arg Arg Glu 290 295 300 Leu Arg Leu Leu Glu Asn Arg Met Ala Glu Lys Gln Glu Glu Glu Arg 305 310 315 320 Leu Arg Cys Arg Asn Met Leu Ser Gln Gly Ala Thr Gln Pro Pro Thr 325 330 335 Glu Pro Pro Ala Glu Pro His Thr Ala Pro Pro Ala Glu Leu Ser Thr 340 345 350 Gly Pro Pro Ala Glu Pro Pro Ala Glu Leu Pro Leu Ser Pro Gly His 355 360 365

Ser Leu Gln His 370

<210> 75 <211> 328

<212> PRT

<213> Bos taurus

<400> 75

Met Asp Ala Thr Leu Leu Leu Asn Val Glu Gly Ile Lys Lys Thr Ile

5 10 15

Leu His Gly Gly Thr Gly Asp Leu Pro Asn Phe Ile Thr Gly Ala Arg 20 25 30

Val Thr Phe His Phe Arg Thr Met Lys Cys Asp Glu Glu Arg Thr Val 35 40 45 UTHou-16UTL final.ST25
Ile Asp Asp Ser Lys Gln Val Gly His Pro Met His Ile Ile Gly
50 55 60 Asn Met Phe Lys Leu Glu Val Trp Glu Ile Leu Leu Thr Ser Met Arg 65 70 75 80 Val Ser Glu Val Ala Glu Phe Trp Cys Asp Thr Ile His Thr Gly Val 85 90 95 Tyr Pro Ile Leu Ser Arg Ser Leu Arg Gln Met Ala Glu Gly Lys Asp 100 105 110 Pro Thr Glu Trp His Val His Thr Cys Gly Leu Ala Asn Met Phe Ala 115 120 125 Tyr His Thr Leu Gly Tyr Glu Asp Leu Asp Glu Leu Gln Lys Glu Pro 130 135 140 Gln Pro Leu Ile Phe Ile Ile Glu Leu Leu Gln Val Glu Ala Pro Ser 145 150 155 160 Gln Tyr Gln Arg Glu Thr Trp Asn Leu Asn Asn Gln Glu Lys Met Gln 165 170 175 Ala Val Pro Ile Leu His Gly Glu Gly Asn Arg Leu Phe Lys Leu Gly 180 185 190 Arg Tyr Glu Glu Ala Ser Asn Lys Tyr Gln Glu Ala Ile Val Cys Leu 195 200 205 Arg Asn Leu Gln Thr Lys Glu Lys Pro Trp Glu Val Gln Trp Leu Lys 210 220 Leu Glu Lys Met Ile Asn Thr Leu Ile Leu Asn Tyr Cys Gln Cys Leu 225 230 235 240 Leu Lys Lys Glu Glu Tyr Tyr Glu Val Leu Glu His Thr Ser Asp Ile 245 250 255 Leu Arg His His Pro Gly Ile Val Lys Ala Tyr Tyr Val Arg Ala Arg 260 265 270 Ala His Ala Glu Val Trp Asn Glu Ala Glu Ala Lys Ala Asp Leu Glu 275 280 285 Lys Val Leu Glu Leu Glu Pro Ser Met Arg Lys Ala Val Gln Arg Glu 290 295 300

Leu Arg Leu Leu Glu Asn Arg Leu Glu Glu Lys Arg Glu Glu Glu Arg 305 310 315 320

Leu Arg Cys Arg Asn Met Leu Gly

<210> 76

<211> 328

<212> PRT

<213> Mus musculus

<220>

<221> peptide

<222> (1)..(328)

<223> Mouse AIPL1 Protein

<400> 76

Met Asp Val Ser Leu Leu Leu Asn Val Glu Gly Val Lys Lys Thr Ile 1 5 10 15

Leu His Gly Gly Thr Gly Glu Leu Pro Asn Phe Ile Thr Gly Ser Arg 20 25 30

Val Thr Phe His Phe Arg Thr Met Lys Cys Asp Glu Glu Arg Thr Val 35 40 45

Ile Asp Asp Ser Lys Gln Val Gly Gln Pro Met Ser Ile Ile Gly
50 55 60

Asn Met Phe Lys Leu Glu Val Trp Glu Thr Leu Leu Thr Ser Met Arg 65 70 75 80

Leu Gly Glu Val Ala Glu Phe Trp Cys Asp Thr Ile His Thr Gly Val 85 90 95

Tyr Pro Met Leu Ser Arg Ser Leu Arg Gln Val Ala Glu Gly Lys Asp 100 105 110

Pro Thr Ser Trp His Val His Thr Cys Gly Leu Ala Asn Met Phe Ala 115 120 125

Tyr His Thr Leu Gly Tyr Glu Asp Leu Asp Glu Leu Gln Lys Glu Pro 130 135 140

Gln Pro Leu Val Phe Leu Tyr Glu Leu Leu Gln Val Glu Ala Pro Asn 145 150 155 160

Glu Tyr Gln Arg Glu Thr Trp Asn Leu Asn Asn Glu Glu Arg Met Gln Page 33 UTHou-16UTL final.ST25 170 175

165

Ala Val Pro Leu Leu His Gly Glu Gly Asn Arg Leu Tyr Lys Leu Gly 180 185 190

Arg Tyr Asp Gln Ala Ala Thr Lys Tyr Gln Glu Ala Ile Val Cys Leu 195 200 205

Arg Asn Leu Gln Thr Lys Glu Lys Pro Trp Glu Val Glu Trp Leu Lys 210 215 220

Leu Glu Lys Met Ile Asn Thr Leu Ile Leu Asn Tyr Cys Gln Cys Leu 225 230 235 240

Leu Lys Lys Glu Glu Tyr Tyr Glu Val Leu Glu His Thr Ser Asp Ile 245 250 255

Leu Arg His His Pro Gly Ile Val Lys Ala Tyr Tyr Met Arg Ala Arg 260 265 270

Ala His Ala Glu Val Trp Asn Ala Glu Glu Ala Lys Ala Asp Leu Glu 275 280 285

Lys Val Leu Glu Leu Glu Pro Ser Met Arg Lys Ala Val Leu Arg Glu 290 295 300

Leu Arg Leu Leu Glu Ser Arg Leu Ala Asp Lys Gln Glu Glu Arg 305 310 315 320

Gln Arg Cys Arg Ser Met Leu Gly 325

<210> 77

<211> 392

<212> PRT

<213> Macaca mulatta

<220>

<221> peptide

<222> (1)..(392)

<223> Rhesus Monkey AILP1 Protein

<400> 77

Met Asp Ala Ala Leu Leu Leu Asn Val Glu Gly Val Lys Lys Thr Ile 1 5 10 15

Leu His Gly Gly Thr Gly Glu Leu Pro Asn Phe Ile Thr Gly Ser Arg 20 25 30

Val Ile Phe His Phe Arg Thr Met Lys Cys Asp Glu Glu Arg Thr Val 35 40 45

Ile Asp Asp Ser Arg Gln Val Asp Gln Pro Met His Ile Ile Gly
50 55 60

Asn Met Phe Lys Leu Glu Val Trp Glu Ile Leu Leu Thr Ser Met Arg 65 70 75 80

Val His Glu Val Ala Glu Phe Trp Cys Asp Thr Ile His Thr Gly Val 85 90 95

Tyr Pro Ile Leu Ser Arg Ser Leu Arg Gln Met Ala Gln Gly Lys Asp 100 105 110

Pro Thr Glu Trp His Val His Thr Cys Gly Leu Ala Asn Met Phe Ala 115 120 125

Tyr His Thr Leu Gly Tyr Glu Asp Leu Asp Glu Leu Gln Lys Glu Pro 130 135 140

Gln Pro Leu Ile Phe Val Ile Glu Leu Leu Gln Val Asp Ala Pro Ser 145 150 155 160

Asp Tyr Gln Arg Glu Thr Trp Asn Leu Ser Asn His Glu Lys Met Lys 165 170 175

Val Val Pro Val Leu His Gly Glu Gly Asn Arg Leu Phe Lys Leu Gly 180 185 190

Arg Tyr Glu Glu Ala Ser Ser Lys Tyr Gln Glu Ala Ile Ile Cys Leu 195 200 205

Arg Asn Leu Gln Thr Lys Glu Lys Pro Trp Glu Val Gln Trp Leu Lys 210 220

Leu Glu Lys Met Ile Asn Thr Leu Thr Leu Asn Tyr Cys Gln Cys Leu 225 230 235 240

Leu Lys Lys Glu Glu Tyr Tyr Glu Val Leu Glu His Thr Ser Asp Ile 245 250 255

Leu Arg His His Pro Gly Ile Val Lys Ala Tyr Tyr Val Arg Ala Arg 260 265 270

Ala His Ala Glu Val Trp Asn Glu Ala Glu Ala Lys Ala Asp Leu Gln 275 280 285 Page 35

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Lys Val Leu Glu Leu Glu Pro Ser Met Gln Lys Ala Val Arg Arg Glu

Leu Arg Leu Leu Glu Asn Arg Met Ala Glu Lys Gln Glu Glu Arg 305 310 315 320

Leu Arg Cys Arg Asn Met Leu Ser Gln Gly Ala Thr Gln Pro Pro Ala 325 330 335

Glu Pro Pro Ala Gln Pro Pro Thr Ala Pro Pro Ala Glu Leu Ser Thr 340 345 350

Gly Pro Pro Ala Asp Pro Pro Ala Glu Pro Pro Thr Ala Pro Pro Ala

Glu Leu Ser Thr Gly Pro Pro Ala Glu Pro Pro Ala Glu Leu Pro Leu 370

Ser Pro Gly His Ser Leu Gln His

78 <210>

<211> 372

<212> **PRT**

Saimiri sciureus

<220>

peptide

<221> <222> (1)..(372)

<223> Squirrel Monkey AIPL1 Protein

<400>

Met Asp Ala Ala Leu Leu Leu Asn Val Glu Gly Val Lys Lys Thr Ile 1 10 15

Leu His Gly Gly Thr Gly Glu Leu Pro Asn Phe Ile Thr Gly Ser Arg
20 25 30

Val Ile Phe His Phe Arg Thr Met Lys Cys Asp Glu Glu Arg Thr Val

Asn Met Phe Lys Leu Glu Val Trp Glu Ile Leu Leu Thr Ser Met Arg 65 70 75 80

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Val Arg Glu Val Ala Glu Phe Trp Cys Asp Thr Ile His Thr Gly Val

85 90 95 Tyr Pro Ile Leu Ser Arg Ser Leu Arg Gln Met Ala Gln Gly Lys Asp 100 105 110 Pro Thr Glu Trp His Val His Thr Cys Gly Leu Ala Asn Met Phe Ala 115 120 125 Tyr His Thr Leu Gly Tyr Glu Asp Leu Asp Glu Leu Gln Lys Glu Pro 130 135 140 Gln Pro Leu Ile Phe Val Ile Glu Leu Leu Gln Val Asp Ala Pro Ser Asp Tyr Gln Arg Glu Thr Trp Asn Leu Ser Asn His Glu Lys Met Lys 165 170 175 Val Val Pro Val Leu His Gly Glu Gly Asn Arg Leu Phe Lys Leu Gly 180 185 190 180 Arg Tyr Glu Glu Ala Ser Ser Lys Tyr Gln Glu Ala Ile Ile Cys Leu 195 200 205 Arg Asn Leu Gln Thr Lys Glu Lys Pro Trp Glu Val Gln Trp Leu Lys 210 225 Leu Glu Lys Met Ile Asn Thr Leu Ile Leu Asn Tyr Cys Gln Cys Leu 225 230 235 240 Leu Lys Lys Glu Glu Tyr Tyr Glu Val Leu Glu His Thr Ser Asp Ile 245 250 255 Leu Arg His His Pro Gly Ile Val Lys Ala Tyr Tyr Val Arg Ala Arg 260 265 270 Ala His Ala Glu Val Trp Asn Glu Ala Glu Ala Lys Ala Asp Leu Gln 275 280 285 Lys Val Leu Glu Leu Glu Pro Ser Met Gln Lys Ala Val Arg Arg Glu 290 295 300 Leu Arg Leu Leu Glu Asn Arg Met Ala Glu Lys Gln Glu Glu Glu Arg 305 310 315 320

Leu Arg Cys Arg Asn Met Leu Ser Gln Gly Ala Thr Trp Ser Pro Ala 325 330 335

Glu Pro Pro Ala Glu Pro Pro Ala Glu Ser Ser Thr Glu Pro Pro Ala 340 345 350

Glu Pro Pro Ala Glu Pro Pro Ala Glu Leu Thr Leu Thr Pro Gly His $355 \hspace{1cm} 360 \hspace{1cm} 365$

Pro Leu Gln His 370